PARTE 2/2

MALATTIE MUSCOLARI
EZIOLOGIA

Forme Geneticamente Determinate

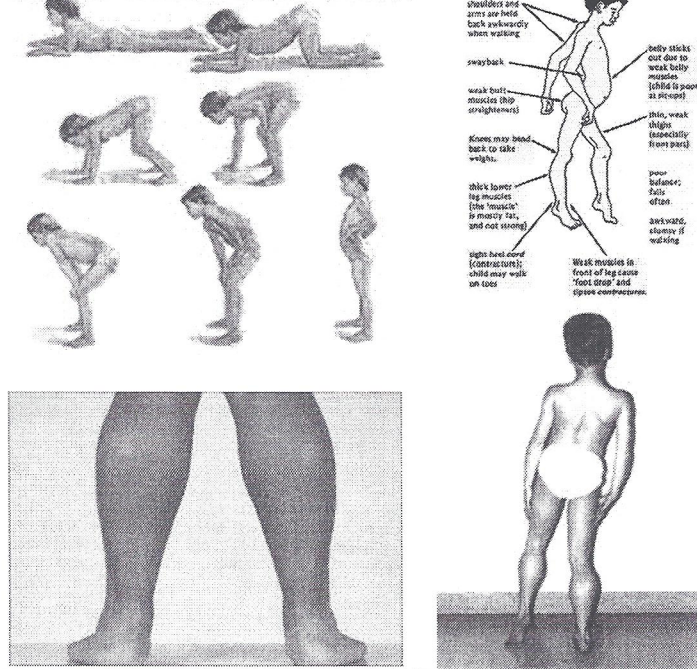
Strutturale	Miopatie Distrofiche Miopatie Congenite	
Metabolica	Glicogenosi Lipidosi Metabolismo Glicidico Metabolismo Purinico Miopatie Mitocondriali	
Canalopatie	Canali K:	Paral Period Ipokaliemica
	Canali Na:	Paral Period Iperkaliemica
	Canali Cl:	Miotonia (non distrofica)
	Canali Ca:	Paral Ipokaliemica Ipertermia Maligna
Miotonia	Congenita Distrofica	

laminin
Collagen
α-dystroglycan
α-SG
β-SG
β-dystro-
glycan
γ-SG
β-SG
SPN
Dystrobrevin
nNOS
DYSTROPHIN
syntrophin
syntrophin
nNOS
actin
cytoskeleton

MALATTIE MUSCOLARI
EZIOLOGIA

Forme Geneticamente Determinate

Muscular Dystrophies (MD)

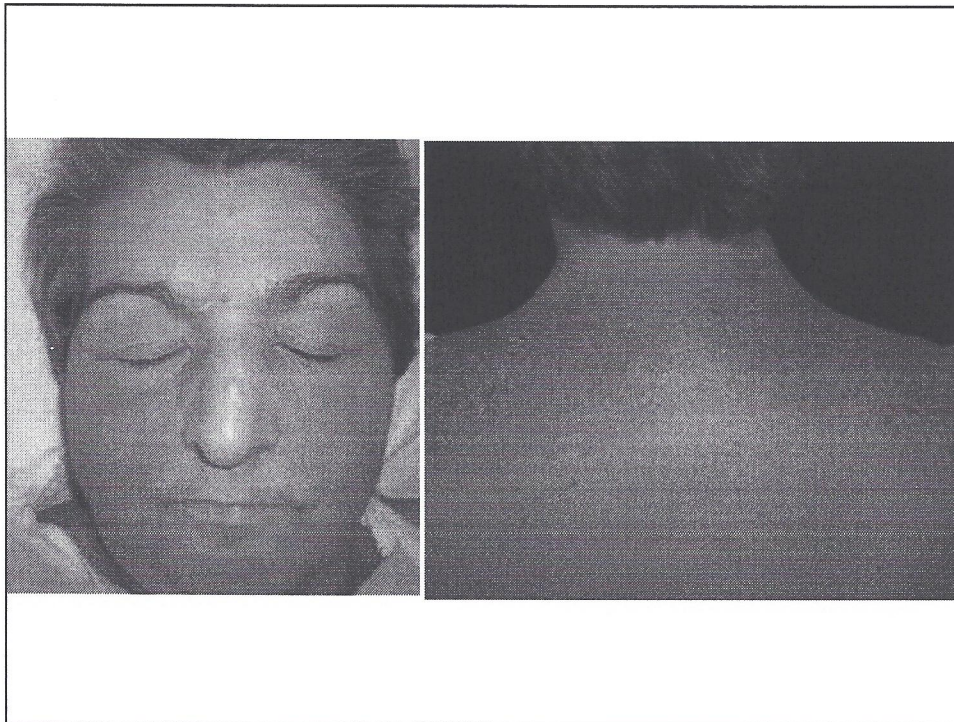
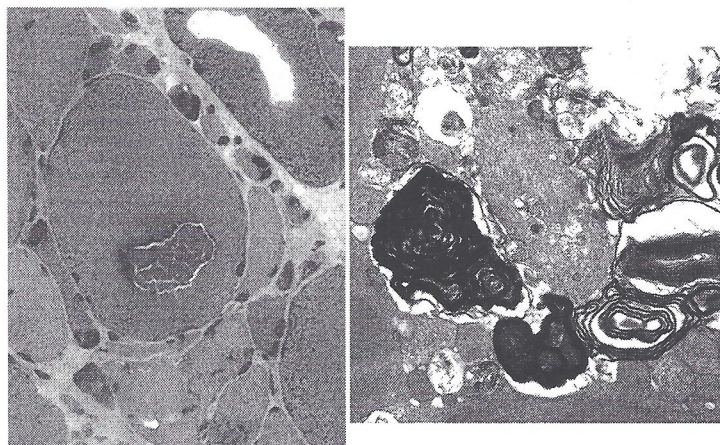
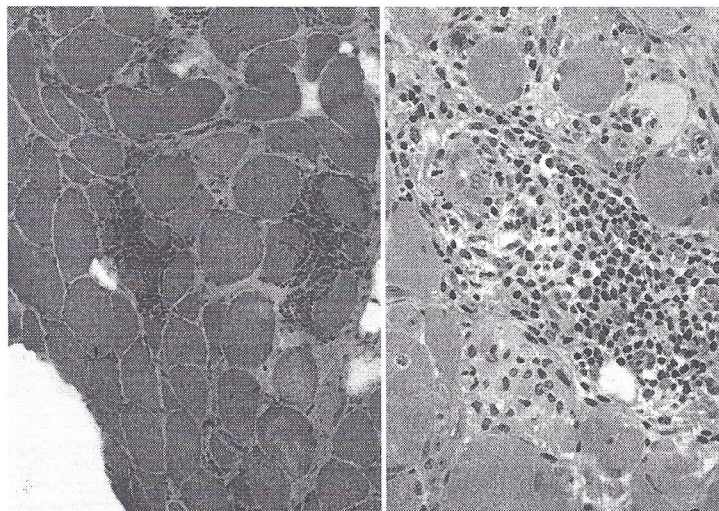
Duchenne MD Becker MD	X-linked; infancy
Limb Girdle MD	AD, AR; 1°-4° decade
Emery-Dreifuss MD	X-linked, AD; childhood, adolescence several variants
Facio-Scapulo-Humeral MD	AD; childhood, early adulthood
Oculo-Pharyngeal MD	AD; late-onset
Myotonic Dystrophy	AD; preschool age 2 forms

MALATTIE MUSCOLARI
EZIOLOGIA

Forme Secondarie

Immuno-mediate	Infiammatoria	Polimiosite Dermatomiosite Miopatia da Corpi Inclusi (IBM)
	Anticorpo-mediata	Miastenia Gravis S miasteniformi
Endocrine	Iper/Ipotiroidismo Iper/Ipoparatiroidismo	
	Ipersteroidismo (sindrome di Cushing)	
Biochimiche	Diabete Mellito Ipokaliemia acquisita Malattie Renali	

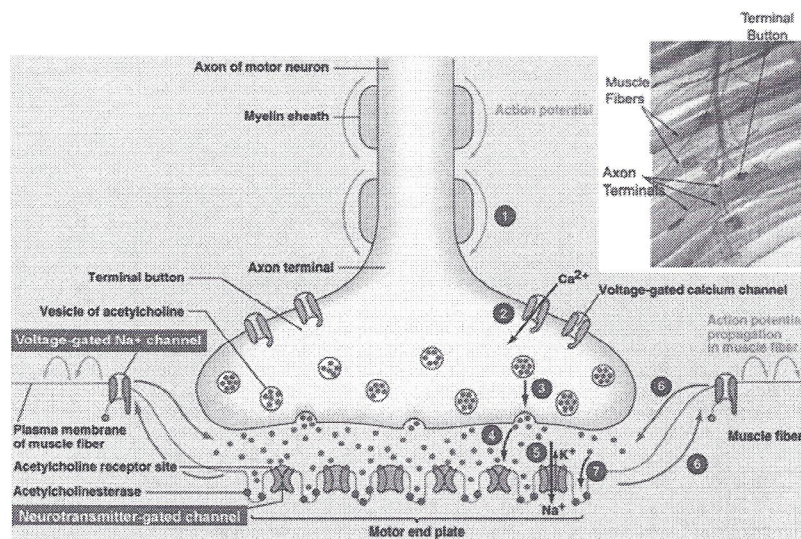
MALATTIE MUSCOLARI
EZIOLOGIA

Forme Secondarie Immuno-Mediate

Infiammatoria	Polimiosite Dermatomiosite Miopatia da Corpi Inclusi (IBM)
Anticorpo-mediata	Miastenia Gravis S miasteniformi

	PM	DM	IBM
Age at onset	>18yrs	Adulthood, childhood	>50yrs
sex	M=F	F>M	M>F
Weakness	proximal	proximal	Proximal, early distal involvement
Familial association	No	No	Yes, in some cases / familial inflammatory myopathies /
Response to treatment	good	better	poor
CTDs	yes	yes	Yes, in up to 20%
malignacy	No	yes, in up to 15% of cases	No
Rash	Absent	Present	Absent
Biopsy	"primary" inflammation with the CD8/MHC-I complex & vacuoles	Perifascicular, perymysial, or privascular infiltrates, perifascicular atrophy	Primary inflammation with CD8/MHC-I complex; vacuolated fibers with b-amyloid deposits , cytochrome oxygenase-negative fibers ; signs of chronic myopathy

MALATTIE MUSCOLARI
EZIOLOGIA

Forme Acquisite

Infettive	virus, parassiti
Tossiche	statine, steroidi, cocaina

The Neuromuscular Junction

MIASTENIA GRAVIS

Clinica

Affaticamento Muscolare (migliora con il riposo)

Tendenza al Reclutamento dei Distretti Muscolari:
coinvolgimento muscolatura facciale: ptosi, disfonia, disfagia

↘

arti, collo…………torace

Diagnostica

Test al Tensilon
EMG (stimolazione ripetitiva)
RMN Torace
Abs anti-Ach-R

Terapia

Farmaci anticolinesterasici
Timectomia
Immunosoppressione: Ig vena
Steroidi
Chemioterapici